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"From genome to function"

This special issue "From genome to function" has been edited by two researchers whose careers have grown along with the rapid developments in this field. In the early days, in the 1980s, one of the goals of human geneticists was to identify DNA polymorphisms, map monogenic human diseases, and identify the culprit genetic mutations and genes responsible for defects. This proved to be an extremely laborious task, requiring multi-million euro projects and large international teams to investigate each disease. One of the dreams at that stage was to speed up the discovery of genetic causes of disease by having a complete human genetic map at hand, so that we could search for genetic loci knowing that we had full genomic coverage. But it was still a tremendous task to move from a genetic locus to gene discovery. Hence, another dream was to have the complete annotated human genome sequence available, so that we could hand pick genes for further analysis without needing complex cloning strategies.

Technological advances have made the challenges of the 1980s reality. We are now witnessing a spectacular increase in the number of loci known to determine monogenic as well as complex human diseases. Soon, we will be able to sequence a human genome for as little as €800 (e.g. the $1000 genome).

This special issue addresses how we can push the boundaries even further, now we have everything we dreamed of 20–30 years ago. One key question is that how can we identify the disease mechanisms now we know the associated genetic loci? With expanding insights, we have started to appreciate the complexity of many of the issues involved. We need to consider not only the coding genes, but also the non-coding genes, and we are now more aware of the role played by "the other genome" in human well-being, i.e. the gut microbiome. This issue has gathered papers on some of the modern technologies and approaches used to identify the genetic variation underlying certain disorders, as well as on the model systems being used to study in depth the mechanisms that translate the genome into function.

Marten Hofker gained his PhD in Medicine at Leiden University (the Netherlands) in 1987, for which he developed a set of X-chromosomal markers that contributed to realizing tests for prenatal diagnosis and

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carrier detection for Duchenne's muscular dystrophy. He did postdoctoral work in the Department of Genetics at the Hospital for Sick Children, Toronto, on the genetic structure of the immunoglobulin heavy chain gene. He later moved to the Department of Human Genetics, Leiden University Medical Center, where he had a personal stipend from the Netherlands Royal Academy of Arts and Sciences (*KNAW*) and an established researcher grant from the Netherlands Heart Foundation to work on the complex genetics of cardiovascular diseases.

In 2000 he became a Professor of Molecular Genetics and head of the respective department at Maastricht University Medical Center (the Netherlands). Since 2007 he has held a similar position at University Medical Center in Groningen. He is well known for his research on metabolism and cardiovascular diseases using mouse models.

Cisca Wijmenga gained her PhD in Medicine at Leiden University (the Netherlands) in 1993, for which she mapped the genetic locus for Facioscapulohumeral muscular dystrophy (FSHD) to chromosome 4q and was able to identify a large deletion of the subtelomeric region as its molecular cause. In 1994 she started postdoctoral work in a new field, working on the role of an inversion of chromosome 16 in acute myeloid leukemia at the National Human Genome Research Institute at NIH. During this period she became interested in the genetics of complex diseases and in 1995 started her own group at Utrecht University to study celiac disease. This work led her to the important observation that several immune-mediated diseases share part of their genetic basis. She is currently investigating the role of non-coding RNAs in immune-mediated diseases and performing more functional genomics studies, funded by a European Research Council advanced grant. Her aim is to translate genetic findings into disease mechanisms. In 2012 she was elected to the Netherlands Royal Academy of Arts and Sciences (*KNAW*) and in 2013 to the Academia Europaea. She has been head of the Department of Genetics at University Medical Center Groningen since 2007.